CarrierSeq ECS Kit: 420-gene panel menu

Increase variant detection rates by targeting a broader range of recessive and inherited disorders

Using next-generation sequencing (NGS), the Ion Torrent™ CarrierSeq™ ECS Kit 420-gene panel targets the full coding region of all genes, enabling the analysis of >36,000 nonbenign ClinVar variants for single-nucleotide variants (SNVs), insertions and deletions (indels), and copy number variants (CNVs).

Disease Research Area	Gene	CNV target
17-beta hydroxysteroid dehydrogenase 3 deficiency	*HSD17B3*	CNV
3-beta-hydroxysteroid dehydrogenase type II deficiency	*HSD3B2*	CNV
3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency	*HMGCL*	CNV
3-methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	CNV
3-methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	CNV
5-alpha reductase deficiency	*SRD5A2*	CNV
6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency	*PTS*	CNV
Abetalipoproteinemia	*MTTP*	CNV
Achalasia-addisonianism-alacrima syndrome	*AAAS*	CNV
Achondrogenesis, type 1B	*SLC26A2*	CNV
Achromatopsia, CNGA3-related	*CNGA3*	CNV
Achromatopsia, CNGB3-related	*CNGB3*	CNV
Acrodermatitis enteropathica	*SLC39A4*	CNV
Acute infantile liver failure, TRMU-related	*TRMU*	CNV
Acyl-CoA oxidase I deficiency	*ACOX1*	CNV
Adrenoleukodystrophy, X-linked	*ABCD1*	CNV
Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome)	*SLC12A6*	CNV

Disease Research Area	Gene	CNV target
Aicardi-Goutieres syndrome, RNASEH2C-related	*RNASEH2C*	CNV
Aicardi-Goutieres syndrome, TREX1-related	*TREX1*	CNV
Aicardi-Goutires syndrome	*SAMHD1*	CNV+
Alkaptonuria	*HGD*	CNV
Alpha-1-antitrypsin deficiency	*SERPINA1*	CNV
Alpha-mannosidosis	*MAN2B1*	CNV
Alpha-thalassemia	*HBA1*	SC
Alpha-thalassemia	*HBA2*	SC
Alpha-thalassemia intellectual disability syndrome, X-linked	*ATRX*	CNV
Alport syndrome, COL4A3-related	*COL4A3*	CNV
Alport syndrome, COL4A4-related	*COL4A4*	CNV
Alport syndrome, X-linked	*COL4A5*	CNV
Alström syndrome	*ALMS1*	CNV
Androgen insensitivity syndrome, X-linked	*AR*	CNV
Argininemia	*ARG1*	CNV
Argininosuccinate lyase deficiency	*ASL*	CNV
Aromatase deficiency	*CYP19A1*	CNV
Arts syndrome, X-linked	*PRPS1*	CNV
Asparagine synthetase deficiency	*ASNS*	CNV
Aspartylglucosaminuria	*AGA*	CNV
Ataxia with vitamin E deficiency	*TTPA*	CNV
Ataxia-telangiectasia	*ATM*	CNV+
Ataxia-telangiectasia-like disorder 1	*MRE11*	CNV
Autism spectrum, epilepsy, and arthrogryposis	*SLC35A3*	CNV
Autoimmune polyendocrinopathy syndrome, type I	*AIRE*	CNV
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	*SACS*	CNV
Bardet-Biedl syndrome 1	*BBS1*	CNV
Bardet-Biedl syndrome 10	*BBS10*	CNV
Bardet-Biedl syndrome 11	*TRIM32*	CNV
Bardet-Biedl syndrome 12	*BBS12*	CNV
Bardet-Biedl syndrome 2	*BBS2*	CNV
Bardet-Biedl syndrome 4	*BBS4*	CNV+
Bardet-Biedl syndrome 6	*MKKS*	CNV
Bardet-Biedl syndrome 9	*BBS9*	CNV
Bare lymphocyte syndrome, CIITA-related	*CIITA*	CNV
Bartter syndrome, type 4a	*BSND*	CNV
Bernard-Soulier syndrome, type A2	*GP1BA*	CNV
Bernard-Soulier syndrome, type B	*GP1BB*	CNV
Bernard-Soulier syndrome, type C	*GP9*	CNV
Beta-hemoglobinopathies	*HBB*	CNV+
Beta-ketothiolase deficiency	*ACAT1*	CNV
Beta-ureidopropionase deficiency	*UPB1*	CNV
Bilateral frontoparietal polymicrogyria	*ADGRG1*	CNV
Biotinidase deficiency	*BTD*	CNV
Bloom syndrome	*BLM*	CNV
Canavan disease	*ASPA*	CNV
Carbamoyl phosphate synthetase I deficiency	*CPS1*	CNV
Carnitine deficiency	*SLC22A5*	CNV
Carnitine palmitoyltransferase IA deficiency	*CPT1A*	CNV

Disease Research Area	Gene	CNV target
Carnitine palmitoyltransferase II deficiency	*CPT2*	CNV
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	CNV
Carpenter syndrome	*RAB23*	CNV
Cartilage-hair hypoplasia	*RMRP*	CNV
Catecholaminergic polymorphic ventricular tachycardia	*CASQ2*	CNV
Cerebrotendinous xanthomatosis	*CYP27A1*	CNV
Ceroid lipofuscinosis, neuronal, 1	*PPT1*	CNV
Ceroid lipofuscinosis, neuronal, 10 (CLN10 disease)	*CTSD*	CNV
Ceroid lipofuscinosis, neuronal, 2	*TPP1*	CNV
Ceroid lipofuscinosis, neuronal, 3	*CLN3*	CNV+
Ceroid lipofuscinosis, neuronal, 5	*CLN5*	CNV
Ceroid lipofuscinosis, neuronal, 6	*CLN6*	CNV
Ceroid lipofuscinosis, neuronal, 7	*MFSD8*	CNV
Ceroid lipofuscinosis, neuronal, 8 (a.k.a. Northern epilepsy)	*CLN8*	CNV
Charcot-Marie-Tooth disease type 4D	*NDRG1*	CNV
Charcot-Marie-Tooth disease with deafness, X-linked	*GJB1*	CNV
Chediak-Higashi syndrome	*LYST*	CNV
Choreo-acanthocytosis	*VPS13A*	CNV+
Choroideremia, X-linked	*CHM*	CNV
Chronic granulomatous disease, CYBA-related	*CYBA*	CNV
Chronic granulomatous disease, X-linked	*CYBB*	CNV
Ciliary dyskinesia, primary 1	*DNAI1*	CNV
Ciliary dyskinesia, primary 3	*DNAH5*	CNV
Ciliary dyskinesia, primary 9	*DNAI2*	CNV
Ciliary dyskinesia, primary, 16	*DNAL1*	CNV
Ciliopathies, RPGRIP1L-related	*RPGRIP1L*	CNV
Citrullinemia, type 1	*ASS1*	CNV
Citrullinemia, type II	*SLC25A13*	CNV
Cockayne syndrome, type A	*ERCC8*	CNV
Cockayne syndrome, type B	*ERCC6*	CNV
Cohen syndrome	*VPS13B*	CNV
Combined malonic and methylmalonic aciduria	*ACSF3*	CNV
Combined oxidative phosphorylation deficiency 1	*GFM1*	CNV
Combined oxidative phosphorylation deficiency 3	*TSFM*	CNV
Combined pituitary hormone deficiency 2	*PROP1*	CNV
Congenital adrenal hyperplasia, 11-beta-hydroxylase-deficient	*CYP11B1*	CNV
Congenital adrenal hyperplasia, 17-alpha-hydroxylase deficiency	*CYP17A1*	CNV
Congenital adrenal hyperplasia, 21-hydroxylase-deficient	*CYP21A2*	SC
Congenital adrenal hypoplasia, X-linked	*NR0B1*	CNV
Congenital amegakaryocytic thrombocytopenia	*MPL*	CNV
Congenital chloride diarrhea	*SLC26A3*	CNV
Congenital disorder of glycosylation, type 1A, PMM2-related	*PMM2*	CNV
Congenital disorder of glycosylation, type 1B	*MPI*	CNV
Congenital disorder of glycosylation, type 1C	*ALG6*	CNV
Congenital Finnish nephrosis	*NPHS1*	CNV
Congenital hyperinsulinism, KCNJ11-related	*KCNJ11*	CNV
Congenital hypothyroidism	*TPO*	CNV
Congenital hypothyroidism	*TSHB*	CNV

Disease Research Area	Gene	CNV target
Congenital insensitivity to pain with anhidrosis (CIPA)	*NTRK1*	CNV
Congenital myasthenic syndrome, CHRNE-related	*CHRNE*	CNV
Congenital myasthenic syndrome, DOK7-related	*DOK7*	CNV
Congenital myasthenic syndrome, RAPSN-related	*RAPSN*	CNV
Congenital neutropenia, HAX1-related	*HAX1*	CNV
Congenital neutropenia, VPS45-related	*VPS45*	CNV
Corneal dystrophy and perceptive deafness	*SLC4A11*	CNV
Corticosterone methyloxidase deficiency	*CYP11B2*	CNV
Costeff syndrome (3-methylglutaconic aciduria, type 3)	*OPA3*	CNV
Creatine transporter defect (cerebral creatine deficiency syndrome 1, X-linked)	*SLC6A8*	CNV
Crigler-Najjar syndrome	*UGT1A1*	CNV
Cystic fibrosis	*CFTR*	CNV+
Cystinosis	*CTNS*	CNV+
Cystinuria, type A	*SLC3A1*	CNV+
Cystinuria, type B	*SLC7A9*	CNV
Cytochrome P450 oxidoreductase deficiency	*POR*	CNV
Cytochrome-c oxidase deficiency	*PET100*	CNV
D-bifunctional protein deficiency	*HSD17B4*	CNV
Deafness, autosomal dominant 36, autosomal recessive 7	*TMC1*	CNV
Deafness, autosomal recessive 16	*STRC*	CNV+
Deafness, autosomal recessive 77	*LOXHD1*	CNV
Deafness, autosomal recessive, 3	*MYO15A*	CNV
Desbuquois dysplasia 1	*CANT1*	CNV
Dihydrolipoamide dehydrogenase deficiency	*DLD*	CNV
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	CNV
Dopa-responsive dystonia	*GCH1*	CNV
Duchenne/Becker muscular dystrophy	*DMD*	CNV+
Dysautonomia, familial (IKBKAP or ELP1)	*IKBKAP*	CNV
Dyskeratosis congenita, RTEL1-related	*RTEL1*	CNV
Dyskeratosis congenita, X-linked	*DKC1*	CNV
Dystrophic epidermolysis bullosa, COL7A1-related	*COL7A1*	CNV
Ehlers-Danlos syndrome, type VIIC	*ADAMTS2*	CNV
Ellis-van Creveld syndrome, EVC-related	*EVC*	CNV
Ellis-van Creveld syndrome, EVC2-related	*EVC2*	CNV
Emery-Dreifuss muscular dystrophy 1, X-linked	*EMD*	CNV
Enhanced S-cone syndrome	*NR2E3*	CNV
Erythrokeratodermia variabilis et progressiva	*GJB3*	CNV
Escobar syndrome	*CHRNG*	CNV
Ethylmalonic encephalopathy	*ETHE1*	CNV
Fabry disease	*GLA*	CNV+
Factor XI deficiency	*F11*	CNV
Familial dilated cardiomyopathy	*TTN*	CNV
Familial hypercholesterolemia, LDLR-related	*LDLR*	CNV
Familial hypercholesterolemia, LDLRAP1-related	*LDLRAP1*	CNV
Familial hyperinsulinism, ABCC8-related	*ABCC8*	CNV
Familial Mediterranean fever	*MEFV*	CNV
Familial nephrogenic diabetes insipidus, AQP2-related	*AQP2*	CNV
Fanconi anemia, group A	*FANCA*	CNV+

Disease Research Area	Gene	CNV target
Fanconi anemia, group C	*FANCC*	CNV+
Fanconi anemia, group G	*FANCG*	CNV
Fanconi anemia, group J	*BRIP1*	CNV
Fibrochondrogenesis, type 2	*COL11A2*	CNV
Fumarase deficiency	*FH*	CNV
Galactokinase deficiency (galactosemia, type II)	*GALK1*	CNV
Galactose epimerase deficiency	*GALE*	CNV
Galactosemia	*GALT*	CNV+
Gaucher disease	*GBA*	CNV
Geroderma osteodysplastica	*GORAB*	CNV
Gitelman syndrome	*SLC12A3*	CNV
Glanzmann thrombasthenia	*ITGB3*	CNV+
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*	CNV
Glutaric acidemia, type 1	*GCDH*	CNV
Glutaric acidemia, type 2A	*ETFA*	CNV
Glutaric acidemia, type 2B	*ETFB*	CNV
Glutaric acidemia, type 2C	*ETFDH*	CNV
Glycine encephalopathy, AMT-related	*AMT*	CNV
Glycine encephalopathy, GLDC-related	*GLDC*	CNV
Glycogen storage disease, type IA	*G6PC*	CNV
Glycogen storage disease, type IB	*SLC37A4*	CNV
Glycogen storage disease, type II (Pompe disease)	*GAA*	CNV+
Glycogen storage disease, type III (Cori/Forbes)	*AGL*	CNV
Glycogen storage disease, type IV	*GBE1*	CNV
Glycogen storage disease, type V (McArdle disease)	*PYGM*	CNV
Glycogen storage disease, type VII	*PFKM*	CNV
GM3 synthase deficiency	*ST3GAL5*	CNV
GRACILE syndrome	*BCS1L*	CNV
Grebe syndrome	*GDF5*	CNV
Guanidinoacetate methyltransferase deficiency	*GAMT*	CNV
Harlequin ichthyosis	*ABCA12*	CNV
Heme oxygenase-1 deficiency	*HMOX1*	CNV
Hemochromatosis, type 1	*HFE*	CNV
Hemochromatosis, type 2A	*HFE2*	CNV
Hemochromatosis, type 3, TFR2-related	*TFR2*	CNV
Hemophilia A	*F8*	CNV
Hemophilia B	*F9*	CNV
Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	*MPV17*	CNV
Hereditary fructose intolerance	*ALDOB*	CNV
Hereditary spastic paraparesis, type 49	*TECPR2*	CNV
Herlitz junctional epidermolysis bullosa, LAMA3-related	*LAMA3*	CNV
Herlitz junctional epidermolysis bullosa, LAMB3-related	*LAMB3*	CNV
Herlitz junctional epidermolysis bullosa, LAMC2-related	*LAMC2*	CNV
Hermansky-Pudlak syndrome 1	*HPS1*	CNV
Hermansky-Pudlak syndrome 3	*HPS3*	CNV
Hermansky-Pudlak syndrome 4	*HPS4*	CNV
Holocarboxylase synthetase deficiency	*HLCS*	CNV
Homocystinuria due to deficiency of MTHFR	*MTHFR*	CNV

Disease Research Area	Gene	CNV target
Homocystinuria, CBS-related	*CBS*	CNV
Homocystinuria, type cblE	*MTRR*	CNV
Hydatidiform mole, recurrent	*NLRP7*	CNV
Hydrolethalus syndrome	*HYLS1*	CNV
Hypermethioninemia	*MAT1A*	CNV
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	*SLC25A15*	CNV
Hyperoxaluria, primary, type 1	*AGXT*	CNV
Hyperphosphatemic familial tumoral calcinosis	*GALNT3*	CNV
Hypohidrotic ectodermal dysplasia	*EDAR*	CNV
Hypohidrotic ectodermal dysplasia, X-linked	*EDA*	CNV
Hypophosphatasia, ALPL-related	*ALPL*	CNV
Hypothyroidism, congenital, nongoitrous, 1	*TSHR*	CNV
Inclusion body myopathy 2	*GNE*	CNV
Infantile neuroaxonal dystrophy 1	*PLA2G6*	CNV
Isolated growth hormone deficiency, type IA/II	*GH1*	CNV+
Isolated growth hormone deficiency, type IB	*GHRHR*	CNV
Isolated growth hormone deficiency, type III, X-linked	*BTK*	CNV
Isovaleric acidemia	*IVD*	CNV
Joubert syndrome 2/Meckel syndrome 2	*TMEM216*	CNV
Juvenile nephronophthisis	*NPHP1*	CNV
Juvenile retinoschisis, X-linked	*RS1*	CNV
Krabbe disease	*GALC*	CNV+
LAMA2-related muscular dystrophy	*LAMA2*	CNV
Lamellar ichthyosis, type 1	*TGM1*	CNV
Leber congenital amaurosis 1	*GUCY2D*	CNV
Leber congenital amaurosis 2	*RPE65*	CNV
Leber congenital amaurosis 8	*CRB1*	CNV
Leber congenital amaurosis, type CEP290	*CEP290*	CNV
Leber congenital amaurosis, type LCA5	*LCA5*	CNV
Leber congenital amaurosis, type RDH12	*RDH12*	CNV
Leigh syndrome	*SURF1*	CNV
Leigh syndrome, French-Canadian type	*LRPPRC*	CNV
Lethal congenital contracture syndrome 1	*GLE1*	CNV
Leukoencephalopathy with vanishing white matter	*EIF2B5*	CNV
Leydig cell hypoplasia	*LHCGR*	CNV
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	CNV
Limb-girdle muscular dystrophy, type 2B	*DYSF*	CNV
Limb-girdle muscular dystrophy, type 2C	*SGCG*	CNV
Limb-girdle muscular dystrophy, type 2D	*SGCA*	CNV
Limb-girdle muscular dystrophy, type 2E	*SGCB*	CNV
Limb-girdle muscular dystrophy, type 2F	*SGCD*	CNV
Limb-girdle muscular dystrophy, type 2I	*FKRP*	CNV
Lipoid congenital adrenal hyperplasia	*STAR*	CNV
Lipoprotein lipase deficiency	*LPL*	CNV
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*	CNV
Lowe syndrome, X-linked	*OCRL*	CNV
Lysinuric protein intolerance	*SLC7A7*	CNV
Lysosomal acid lipase deficiency	*LIPA*	CNV

Disease Research Area	Gene	CNV target
Malonyl-CoA decarboxylase deficiency	*MLYCD*	CNV
Maple syrup urine disease, type 1A	*BCKDHA*	CNV
Maple syrup urine disease, type 1B	*BCKDHB*	CNV
Maple syrup urine disease, type 2	*DBT*	CNV
Meckel-Gruber syndrome, type 1	*MKS1*	CNV
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*	CNV
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*	CNV
Megaloblastic anemia syndrome	*SLC19A2*	CNV
Menkes syndrome, X-linked	*ATP7A*	CNV
Mental retardation, autosomal recessive 3	*CC2D1A*	CNV+
Mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, and keratoderma (MEDNIK)	*AP1S1*	CNV
Metachromatic leukodystrophy, ARSA-related	*ARSA*	CNV
Metachromatic leukodystrophy, PSAP-related	*PSAP*	CNV
Methylmalonic aciduria and homocystinuria, type cblC	*MMACHC*	CNV
Methylmalonic aciduria and homocystinuria, type cblD	*MMADHC*	CNV
Methylmalonic aciduria, MMAA-related	*MMAA*	CNV
Methylmalonic aciduria, MMAB-related	*MMAB*	CNV
Methylmalonic aciduria, type mut(0)	*MUT*	CNV
Microcephaly, postnatal progressive, with seizures and brain atrophy	*MED17*	CNV
Microphthalmia/Anophthalmia, VSX2-related	*VSX2*	CNV
MIRAGE syndrome	*SAMD9*	CNV
Mitochondrial complex I deficiency	*NDUFS4*	CNV
Mitochondrial complex I deficiency, ACAD9-related	*ACAD9*	CNV
Mitochondrial complex I deficiency, NDUFAF5-related	*NDUFAF5*	CNV
Mitochondrial complex I deficiency, NDUFS6-related	*NDUFS6*	CNV
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	*SUCLA2*	CNV
Mitochondrial myopathy and sideroblastic anemia (MLASA1)	*PUS1*	CNV
Molybdenum cofactor deficiency	*MOCS1*	CNV
Mucolipidosis II/IIIA	*GNPTAB*	CNV
Mucolipidosis III gamma	*GNPTG*	CNV
Mucolipidosis, type IV	*MCOLN1*	CNV+
Mucopolysaccharidosis, type I (Hurler syndrome)	*IDUA*	CNV
Mucopolysaccharidosis, type II (Hunter syndrome)	*IDS*	CNV
Mucopolysaccharidosis, type IIIA (Sanfilippo A)	*SGSH*	CNV
Mucopolysaccharidosis, type IIIB (Sanfilippo B)	*NAGLU*	CNV
Mucopolysaccharidosis, type IIIC (Sanfilippo C)	*HGSNAT*	CNV
Mucopolysaccharidosis, type IIID (Sanfilippo D)	*GNS*	CNV
Mucopolysaccharidosis, type IVA	*GALNS*	CNV
Mucopolysaccharidosis, type IVB / GM1 gangliosidosis	*GLB1*	CNV
Mucopolysaccharidosis, type VI (Maroteaux-Lamy)	*ARSB*	CNV
Mucopolysaccharidosis, type VII	*GUSB*	CNV
Mulibrey nanism syndrome	*TRIM37*	CNV
Multiple congenital anomalies-hypotonia-seizures syndrome 1	*PIGN*	CNV
Multiple sulfatase deficiency	*SUMF1*	CNV
Muscle-eye-brain disease, POMGNT1-related	*POMGNT1*	CNV
Myasthenic syndrome, congenital, 22	*PREPL*	CNV+
Myoneurogastrointestinal encephalopathy (MNGIE)	*TYMP*	CNV

Disease Research Area	Gene	CNV target
Myotubular myopathy, X-linked	*MTM1*	CNV
N-acetylglutamate synthase deficiency	*NAGS*	CNV
Nemaline myopathy, NEB-related	*NEB*	CNV+
Niemann-Pick disease, type C1/D	*NPC1*	CNV
Niemann-Pick disease, type C2	*NPC2*	CNV
Niemann-Pick disease, types A/B	*SMPD1*	CNV
Nijmegen breakage syndrome	*NBN*	CNV
Non-syndromic hearing loss (a.k.a. connexin 26)	*GJB2*	CNV+
Non-syndromic hearing loss (a.k.a. connexin 30)	*GJB6*	CNV+
Oculocutaneous albinism, type 1	*TYR*	CNV
Oculocutaneous albinism, type 3	*TYRP1*	CNV
Oculocutaneous albinism, type 4	*SLC45A2*	CNV
Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge syndrome	*WNT10A*	CNV
Omenn syndrome, RAG1-related	*RAG1*	CNV
Omenn syndrome, RAG2-related	*RAG2*	CNV
Ornithine aminotransferase deficiency	*OAT*	CNV
Ornithine transcarbamylase deficiency	*OTC*	CNV
Osteopetrosis, infantile malignant, TCIRG1-related	*TCIRG1*	CNV
Pantothenate kinase-associated neurodegeneration	*PANK2*	CNV
Papillon-Lefevre syndrome	*CTSC*	CNV
Pendred syndrome	*SLC26A4*	CNV
Peroxisome biogenesis disorder 1A (Zellweger)	*PEX1*	CNV
Peroxisome biogenesis disorder 3A (Zellweger)	*PEX12*	CNV
Peroxisome biogenesis disorder 4A (Zellweger)	*PEX6*	CNV
Peroxisome biogenesis disorder 5A (Zellweger)	*PEX2*	CNV
Peroxisome biogenesis disorder 6A (Zellweger)	*PEX10*	CNV
Persistent Müllerian duct syndrome, type 1	*AMH*	CNV
Persistent Müllerian duct syndrome, type 2	*AMHR2*	CNV
Phenylketonuria	*PAH*	CNV+
Phosphoglycerate dehydrogenase deficiency	*PHGDH*	CNV
POLG-related disorders	*POLG*	CNV
Polycystic kidney disease, autosomal recessive	*PKHD1*	CNV
Pontocerebellar hypoplasia	*TSEN54*	CNV
Pontocerebellar hypoplasia, type 1 and 6, RARS2-related	*RARS2*	CNV
Pontocerebellar hypoplasia, type 1A	*VRK1*	CNV
Pontocerebellar hypoplasia, type 1B	*EXOSC3*	CNV
Pontocerebellar hypoplasia, type 2D	*SEPSECS*	CNV
Pontocerebellar hypoplasia, type 2E	*VPS53*	CNV
Primary congenital glaucoma	*CYP1B1*	CNV
Primary hyperoxaluria, type 2	*GRHPR*	CNV
Primary hyperoxaluria, type 3	*HOGA1*	CNV
Progressive familial intrahepatic cholestasis, type 1	*ATP8B1*	CNV
Progressive familial intrahepatic cholestasis, type 2	*ABCB11*	CNV
Progressive familial intrahepatic cholestasis, type 3	*ABCB4*	CNV
Progressive pseudorheumatoid dysplasia	*WISP3*	CNV
Prolidase deficiency	*PEPD*	CNV
Propionic acidemia, PCCA-related	*PCCA*	CNV
Propionic acidemia, PCCB-related	*PCCB*	CNV

Disease Research Area	Gene	CNV target
Prothrombin deficiency	*F2*	CNV
Pseudocholinesterase deficiency	*BCHE*	CNV
Pseudoxanthoma elasticum	*ABCC6*	CNV
Pycnodysostosis	*CTSK*	CNV
Pyridoxal 5' -phosphate-dependent epilepsy	*PNPO*	CNV
Pyridoxine-dependent epilepsy	*ALDH7A1*	CNV
Pyruvate carboxylase deficiency	*PC*	CNV
Pyruvate dehydrogenase deficiency, PDHB-related	*PDHB*	CNV
Pyruvate dehydrogenase deficiency, X-linked	*PDHA1*	CNV
Renal tubular acidosis and deafness, ATP6V1B1-related	*ATP6V1B1*	CNV
Retinal dystrophies, RLBP1-associated	*RLBP1*	CNV
Retinitis pigmentosa 25	*EYS*	CNV
Retinitis pigmentosa 26	*CERKL*	CNV
Retinitis pigmentosa 28	*FAM161A*	CNV
Retinitis pigmentosa 59	*DHDDS*	CNV
RETT syndrome	*MECP2*	CNV
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*	CNV
Rhizomelic chondrodysplasia punctata, type 3	*AGPS*	CNV
Roberts syndrome	*ESCO2*	CNV
Salla disease	*SLC17A5*	CNV
Sandhoff disease	*HEXB*	CNV
Schimke immunoosseous dysplasia	*SMARCAL1*	CNV
Segawa syndrome, TH-related	*TH*	CNV
Severe combined immunodeficiency, ADA-related	*ADA*	CNV
Severe combined immunodeficiency, type athabaskan	*DCLRE1C*	CNV
Severe combined immunodeficiency, X-linked	*IL2RG*	CNV
Short chain acyl-CoA dehydrogenase deficiency	*ACADS*	CNV
Short/branched chain acyl-CoA dehydrogenase deficiency	*ACADSB*	CNV
Shwachman-Diamond syndrome	*SBDS*	CNV
Sialidosis	*NEU1*	CNV
Sjögren-Larsson syndrome	*ALDH3A2*	CNV
Smith-Lemli-Opitz syndrome	*DHCR7*	CNV
Spastic paraplegia type 15	*ZFYVE26*	CNV
Spinal muscular atrophy	*SMN1*	SC
Spondylothoracic dysostosis, MESP2-related	*MESP2*	CNV
Stargardt disease, type 1	*ABCA4*	CNV
Steroid-resistant nephrotic syndrome	*NPHS2*	CNV
Stuve-Wiedemann syndrome	*LIFR*	CNV
Tay-Sachs disease	*HEXA*	CNV+
Tricho-hepato-enteric syndrome	*TTC37*	CNV
Trifunctional protein deficiency	*HADHB*	CNV
Tyrosinemia, type 3	*HPD*	CNV
Tyrosinemia, type I	*FAH*	CNV
Tyrosinemia, type II	*TAT*	CNV
Usher syndrome, type 1B	*MYO7A*	CNV
Usher syndrome, type 1C	*USH1C*	CNV
Usher syndrome, type 1D	*CDH23*	CNV
Usher syndrome, type 1F	*PCDH15*	CNV+

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Disease Research Area	Gene	CNV target
Usher syndrome, type 2A	*USH2A*	CNV+
Usher syndrome, type 3	*CLRN1*	CNV
Very long chain acyl-CoA dehydrogenase deficiency	*ACADVL*	CNV
Vitamin D–dependent rickets, type 1A	*CYP27B1*	CNV
Von Willebrand disease	*VWF*	CNV
Walker-Warburg syndrome, FKTN-related	*FKTN*	CNV
Werner syndrome	*WRN*	CNV
Wilson disease	*ATP7B*	CNV
Wiskott-Aldrich syndrome, X-linked	*WAS*	CNV
Wolcott-Rallison syndrome	*EIF2AK3*	CNV
Woolly hair/hypotrichosis syndrome	*LIPH*	CNV
Xeroderma pigmentosum group A	*XPA*	CNV
Xeroderma pigmentosum group C	*XPC*	CNV
Xeroderma pigmentosum group E	*DDB2*	CNV
Xeroderma pigmentosum variant	*POLH*	CNV
Xeroderma pigmentosum, group B	*ERCC3*	CNV
Xeroderma pigmentosum, group D	*ERCC2*	CNV
Xeroderma pigmentosum, group F	*ERCC4*	CNV
Xeroderma pigmentosum, group G	*ERCC5*	CNV

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